CRANIOSTENOSIS WITH FAMILIAL VITAMIN-D-RESISTANT RICKETS

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The syndrome of rickets resistant to normal therapeutic doses of vitamin D but amenable to treatment with massive doses (up to 1,500,000 i.u. daily) was first described by Albright, Butler, and Bloomberg (1937). Since then the literature has become extensive, and it is now recognized that the condition is by no means rare. Holt (1950), Scott (1951), and Pedersen and McCarroll (1951) give good accounts of it; the latter, in the course of an account of 25 cases, point out the strongly familial nature of the disease. Albright *et al.* established by metabolic studies that there is a resistance to normal doses of vitamin D not attributable to malabsorption of the vitamin; and Robertson, Harris, and McCune (1942) attribute the complaint to deficiency of an enzyme, of which vitamin D is said to be the prosthetic group, normally responsible for absorption of phosphate by the gut and its retention by the kidney. Freeman and Dunsky (1950), however, failed to get a restoration of normal serum phosphate despite successful therapy, and they question whether vitamin-D treatment corrects the metabolic defect.

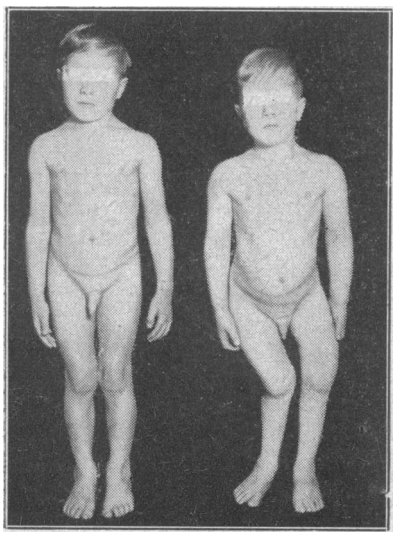

FIG. 1.—The patient aged 6¾ (right), with his brother aged 5½.

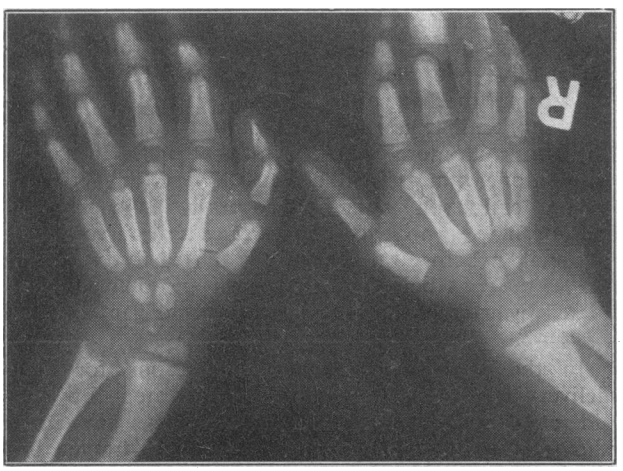

FIG. 2.—Active rickets (aged 3).

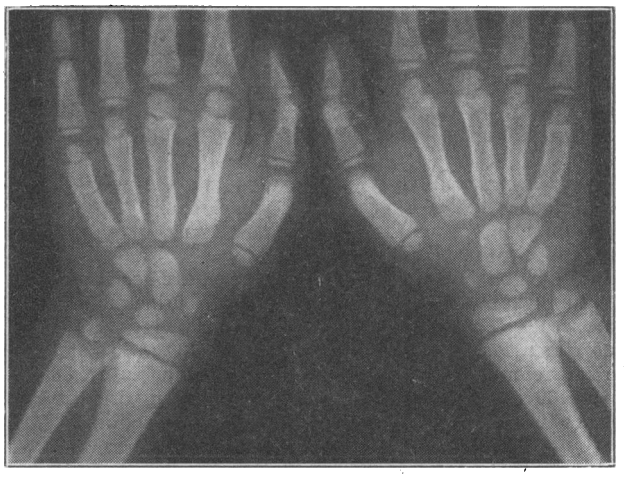

FIG. 3.—Improvement (aged 6¾).

The deformities occurring in vitamin-D-resistant rickets are similar to those of classical rickets; indeed, there is no reason to suppose that the mechanism of deformity is in any way different in the two conditions. In one respect, however, there appears to be a difference—that in the familial resistant variety craniostenosis occasionally occurs. This has been stated only once in the literature, by Imerslund (1951), who describes a boy with typical vitamin-D-resistant rickets whose mother has old rachitic deformities. This child has a considerable degree of scaphocephaly following on premature fusion of the sagittal suture, and necessitating craniotomy for the relief of increased intracranial pressure.

This is not the only occasion on which scaphocephaly has been reported, for it is evident in an illustration of one of the cases of Albright *et al.*, and Pedersen and McCarroll mention the deformity while attributing it to rickets. In addition, Scott comments on the existence, in some cases, of frontal bossing with increased antero-posterior skull diameter and decreased transverse measurement. None of these authors, however, make mention of craniostenosis due to premature fusion of the sagittal suture, which clearly occurred in Imerslund's patient, whose mother also showed a small degree of scaphocephaly. No attempt has been made to explain this association of conditions, the one manifested as delay in ossification, the other by its premature occurrence. We regard this association as unusual enough to warrant the following description of a second case showing both phenomena.

Case Report

A boy aged 6 was first diagnosed as suffering from classical rickets when 2–3 years old. He is the eldest of three, the third child having died of meningitis in infancy. Fig. 1 shows the patient with his younger brother. All three were born by caesarean section, as the mother has old rickets with a contracted pelvis and genu valgum. She has no obvious biochemical abnormality apart from the rather low figure of 1.7 mg. of phosphorus per 100 ml. for her plasma inorganic phosphate. She has not got scaphocephaly. No relevant family history was obtained, as the family is not particularly "clannish" and little is known of some relatives. The patient had a normal diet as an infant and an adequate supplement of national cod-liver oil—2 dr. (7 ml.) daily from 1 year onwards. Despite this he developed frank rickets when 2 years old. A photograph (too "grainy" for repro-

duction) at 20 months shows him walking normally, but with the skull deformity already apparent. At 3 years he had active rickets (Fig. 2). He was given routine anti-rachitic treatment (sunlight and fish-liver oils). When first seen by us in August, 1952, at 6¾ years, his wrists were as shown in Fig. 3. His skull deformity is shown in Figs. 4 and 5.

Biochemical Investigations (in each case the mean of two or more estimates is given).—Serum calcium, 10.2 mg. per 100 ml.; plasma inorganic phosphate, 2 mg. of phosphorus per 100 ml.; alkaline phosphatase, 31 King–Armstrong units; blood urea, 26 mg. per 100 ml.; urea-clearance test (Van Slyke), 120% of average normal; urine, N.A.D. on numerous occasions. In addition the following were found on one occasion each: plasma chloride, 100 mEq per litre; alkali reserve, 27 mEq per litre; fasting blood sugar, 88 mg. per 100 ml.; faecal fat, 15.7 g. per 100 g. dried faeces (unsplit, 1.4 g.); three-day fat balance, absorption 94%.

After treatment with 50,000 i.u. of vitamin D daily by mouth, starting in early September, 1952, the serum calcium rose slightly to 11.2 mg. and the plasma inorganic phosphate rose progressively to 2.7 mg. after three weeks and to 4.3 mg. in six weeks, but fell to 2.4 mg. after 25 weeks of the same treatment. The alkaline phosphatase did not vary significantly over this period. The only definite biochemical abnormalities were the low plasma inorganic phosphate and the slightly raised phosphatase. The former responded temporarily to the relatively small dose of vitamin D. Renal function remained normal throughout.

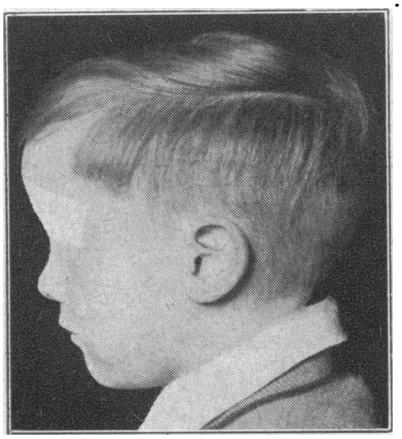

FIG. 4.—The patient aged 6½.

We are indebted to Dr. C. H. Wood, for the following report: "November, 1948:—Generalized decalcification with bowing of long bones; widening of the space between epiphyses and metaphyses; saucer-shaped and irregular metaphyses; these changes are more advanced in the legs. Skull: marked scaphocephaly with closure of the sagittal suture. August, 1952:—Appreciable improvement since earlier films, although not healed. February, 1953:—Improvement maintained, but still not healed; *x*-ray films of patient's mother showed typical changes of healed rickets but no scaphocephaly."

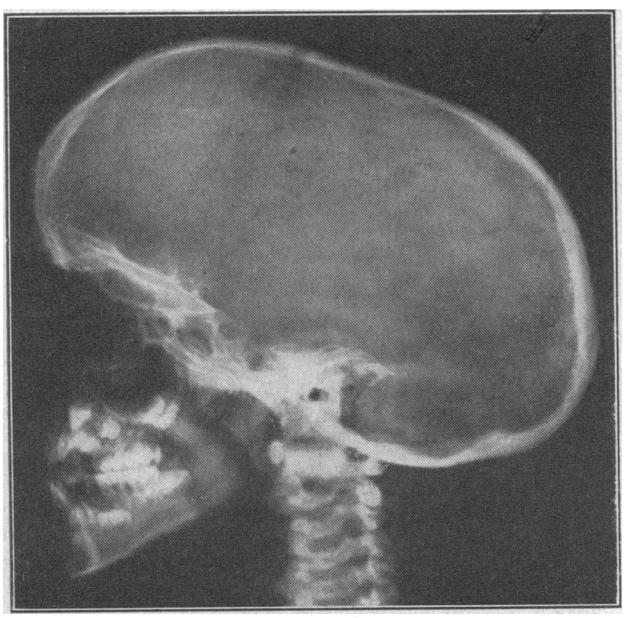

FIG. 5.—Skull (aged 6½).

There seemed to be little doubt of the diagnosis of vitamin-D-resistant rickets, and more vigorous therapy was instituted. In addition he has well-marked craniostenosis from premature fusion of his sagittal suture, a condition only once previously described in association with resistant rickets.

Summary

The second reported case of craniostenosis associated with vitamin-D-resistant rickets is presented together with evidence to suggest that the association of these conditions may have occurred previously and been mistaken for a purely rachitic phenomenon.

We are indebted to Dr. J. M. Macfie for permission to publish this case; to Dr. C. H. Wood for the *x*-ray films; and to the department of clinical photography, General Hospital, Nottingham, for the other illustrations.

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ANAESTHESIA IN HOSPITAL BREECH DELIVERY

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For practical purposes an infant may be delivered by the breech in one of two very different ways: an assisted breech delivery or breech extraction. The two essential features of the former type of delivery are that labour should be left to the natural forces so far as possible and that any intervention should imitate the normal mechanism of breech delivery. In practice this implies that assistance is confined to the birth of the arms and, more especially, of the head. In contradistinction to an assisted breech delivery, delivery in a breech extraction is entirely operative, the maternal powers playing no part. In most cases intervention is undertaken before the breech has started to distend the vulva, the legs being brought down and delivery completed by a combination of traction and gentle fundal pressure.

In the absence of definite indications for early delivery, choice of methods is dictated by consideration of the foetal hazards. These have been shown to be due to two factors—namely, trauma and anoxia, of which the former is by far the more